MEDIA RELEASE

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Today HCU Network America announced the recipient of their first newborn screening research grant. The New England Newborn Screening Program, an initiative of UMass Chan Medical School's Commonwealth Medicine division, received the award to explore the development of reference ranges for additional newborn screening markers for early detection of classical homocystinuria and remethylation disorders. The research, led by Devinder Kaur, PhD, assistant professor of pediatrics at UMass Chan, aims to establish normal reference ranges for total homocysteine, along with other analytes collected by healthy newborns during the 24-48 newborn screening period. This will support the development of algorithms that will incorporate information on a variety of other variables in the future. Dr. Kaur, who is leading the research, joined the New England Newborn Screening Program in 2017 as a senior scientist.

Homocystinuria is a group of rare metabolic genetic disorders, of which all have high homocysteine. There are approximately 10 conditions that fall under the term homocystinuria. Under the United States federal Recommended Uniform Screening Panel (RUSP), the only type of homocystinuria recommended for newborn screening is classical homocystinuria, but a majority also will screen for cobalamin C - other states may pick up other types, but it's not ensured.

Massachusetts led the way with newborn screening (NBS) for classical HCU by adding a Guthrie bacterial inhibition assay for methionine (MET) to their NBS panel in 1968. Slowly many other states joined the ranks, but it wasn't until 2009, after the Newborn Screening Saves Lives Act of 2007, that classical HCU officially became part of the Recommended Uniform Screening Panel (RUSP). Between 1968 and 2009 many changes took place in NBS, and tandem mass spectrometry which includes MET became the new standard for screening.

Unfortunately, though, MET alone is not as sensitive a biomarker. In many cases, the baby who has classical HCU has not had an increased level of MET or a level sufficiently increased to be identified at a time of 24-48 hours of age, when the newborn screening blood specimen is collected. In addition, there is also no harmonized cut-off level for increased MET. Every state sets their own cut-offs, which range from 35 umol/L (hereinafter referred to as uM) to 100 uM across the US. There are known cases of HCU with just a mild elevation of MET who were detected in NBS because the program had a relatively low cut-off but would have been missed if born in a state with a higher cut-off. Higher MET cutoff levels are often set to minimize the number of false positives, but, unfortunately, are likely to lead to missing HCU.

The proposed study will help to determine distribution of four HCU specific markers in the healthy standard newborn population and to determine appropriate cut-offs.

Dr. Kaur, who is leading the research says “I am extremely honored and happy to receive this grant from HCU Network America, as this builds on our previous and current work to improve and expand screening methods for the homocystinurias using tandem mass spectrometry with the ultimate goal to help prevent treatable disorders through early detection. This project will leverage the New England Newborn Screening Program’s technical and clinical expertise along with the UMass Chan infrastructure that provides access to screening for a wide variety of disorders; by identifying markers, developing assays, and by generating data, algorithms and protocols that could be adopted nationwide in screening for medically relevant conditions. On behalf of the New England Newborn Screening team, I would like to express my sincere gratitude to HCU Network America for their generous support”.

Dr. Harvey Levy, a newborn screening pioneer and HCU Network America board member said, “We are pleased to fund this grant to develop reference ranges for analytes measured in newborn screening that are markers for the homocystinurias and remethylation disorders, which will further support the adoption of two tier testing to improve the sensitivity of newborn screening and enable these infants to be diagnosed at birth and given the best chance of leading healthy and productive lives”,

The funding from HCU Network America is through the HCU Network America’s Research Fund and the Hempling Foundation for Homocystinuria Research, established in memory of Judy and Susie Hempling, two young girls from Buffalo, NY whose lives were cut short by HCU in the 1970s. HCU Network America would also like to thank their community for their fundraising efforts that helped contribute to making this grant possible.

About HCU Network America:

HCU Network America is a 501c (3) non-profit organization founded in 2016 dedicated to helping patients and their families affected by Homocystinuria (HCU) and related disorders. The mission of the organization is to inform and provide resources for patients and families, create connections, influence state and federal policy, and support advancement of diagnosis and treatment for HCU and related disorders.